

Table 2. Summary of recommendations.

System	Evaluation	When	Management and recommendations
Ocular	Follow-up with ophthalmologist	At diagnosis	<ul style="list-style-type: none"> • Vision screening to monitor for refractive errors and strabismus • Strabismus may require patching or surgery
Ears, nose, throat (ENT)	Follow-up with otolaryngology (ENT)	As needed	<ul style="list-style-type: none"> • Hearing screening • Monitoring recurrent ear infections and epistaxis • Standard surgical management as required
Dental	Ongoing follow-up with dentist for abnormal enamel, tooth shape, and number	Ongoing	<ul style="list-style-type: none"> • Initial pediatric dental evaluation by 1 year of age • May need increased frequency of checkups and cleanings • Dental care may require assistance with daily brushing and flossing
Cardiovascular	Evaluation by cardiologist	At diagnosis	<ul style="list-style-type: none"> • Echocardiogram • Evaluation in infancy for evidence of congenital cardiovascular disease • Referral to cardiologist and/or cardiothoracic surgeon as needed
Gastrointestinal	Consider referral to gastroenterologist and/or feeding specialist or nutritionist	As needed	<ul style="list-style-type: none"> • Consider evaluation for feeding and growth as indicated • Behavioral and/or medical treatment of constipation, if persistent • Age-specific treatment for reflux, including testing for food allergies
Renal	Follow-up with urologist	As needed	<ul style="list-style-type: none"> • Consider evaluation for enuresis, if persistent • Consider behavioral interventions, including alarm techniques, if indicated • May require evaluation of medications that could contribute to enuresis
Musculoskeletal	Referral to orthopedist as needed	As needed	<ul style="list-style-type: none"> • Evaluation for chest anomalies and flat feet • Routine screening for scoliosis
Pulmonary and sleep	Referral to pulmonologist/sleep clinic, as needed	As needed	<ul style="list-style-type: none"> • Sleep study, as needed • Recommendations for implementing healthy sleep hygiene habits • Management of sleep disturbance, as needed
Allergy and immunology	Referral to specialist as needed	As needed	<ul style="list-style-type: none"> • Allergy testing, as needed • Standard management for asthma, allergies, and eczema • Assess for food allergies
Neurological	Referral to neurologist, as needed	At diagnosis and ongoing	<ul style="list-style-type: none"> • Evaluation for seizures, if indicated • Evaluation of muscle tone, if indicated
Neurodevelopmental	Referral to developmental pediatrician and/or clinical psychologist	Follow-up throughout early childhood	<ul style="list-style-type: none"> • Evaluation for autism spectrum disorder, cognitive ability, executive function deficits • Evaluation for developmental needs and early intervention (e.g., physical therapy, speech–language therapy, cognitive behavioral therapy for social skills training) • Evaluation of fine motor function and intervention (e.g., occupational therapy) • Adaptive behavior (e.g., social skills training)
Psychiatric	Referral to child/adult psychiatrist	Childhood to early adult	<ul style="list-style-type: none"> • Periodic evaluation for anxiety disorders, attention deficit hyperactivity disorder, emerging features of prodrome/psychosis • Cognitive behavioral therapy for anxiety • Medications for anxiety, attention deficit–hyperactivity disorder (ADHD), psychosis may be indicated

DISCUSSION

Here we report comprehensive descriptions of 3q29 deletion syndrome, by direct systematic evaluation of 32 study subjects using a defined protocol and gold-standard instruments. While nearly all major systems are affected, the ocular, dental, cardiovascular, gastrointestinal, renal, musculoskeletal, and neurologic systems should be prioritized for evaluation and follow-up.

There is also a significant burden of neurodevelopmental and neuropsychiatric illness, requiring intervention and support across the lifespan. Our findings suggest recommendations for clinical care, described in detail below and summarized in Table 2.

Almost 60% of subjects reported ocular manifestations; the most frequent were strabismus and refractive errors, including myopia and astigmatism. In some cases, the strabismus required